GeneDx to Present Data at the 2024 American College of Medical Genetics (ACMG) Annual Meeting Demonstrating Clinical Superiority of its Exome, Paving the Way for the Future of Genomics

March 14, 2024

STAMFORD, Conn., March 14, 2024 (GLOBE NEWSWIRE) -- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced its scientific contributions at the 2024 American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting. Across one platform presentation, an industry symposium and six poster presentations, including one selected as a “Rapid Fire Poster Session,” an invitation-only presentation of the top 20 ACMG posters, GeneDx continues to demonstrate its industry leadership in shaping the future landscape of genomics.

One of the GeneDx poster sessions includes data demonstrating the ability of exome sequencing to detect small copy number variants (CNVs) below the reporting threshold of chromosomal microarray (CMA) and the detection of CNVs at nearly 99% concordance with CMA. These findings represent improved data quality from pipeline and platform improvements. Combined with its ability to identify many other types of variants compared to CMA and its nearly three times higher diagnostic yield,[1,2,3] these findings demonstrate the clinical superiority of an exome over CMA. These data support the current clinical guidelines recommending exome as a first-tier test and represent GeneDx’s groundbreaking approach to genomic data analysis and interpretation, aimed at eliminating the need for additional tests when ordering exome first.

In its platform presentation, GeneDx will highlight its newly developed statistical approach for predicting the impact of many missense variants with a precision of 99%. This proof-of-concept research has the potential to decrease the variants of unknown significance (VUS) rate and drive greater operational efficiency and diagnostic precision by decreasing the number of variants requiring additional vetting.

In addition to its technological and scientific advancements, GeneDx is committed to democratizing access to genomic testing by eliminating barriers and unlocking the bottlenecks of care. Through an industry-sponsored session, GeneDx will explore the gap between the availability of the medical genetics workforce and the genetic testing needs of pediatric patients. This session features a discussion comparing viewpoints and opportunities around non-geneticists ordering genomic sequencing as part of routine care, with the goal of improving patient outcomes.

"We are motivated to continue to push the boundaries of genomics to transform the future of healthcare," said Paul Kruszka, Chief Medical Officer at GeneDx. "Through our relentless commitment, we are revolutionizing genetic testing and diagnosis, and empowering healthcare providers to deliver personalized care to even more patients."

Full 2024 ACMG Conference Participation:

Industry Platform Presentation: Thursday, 3/14/2024, 11:30 am - 12:45 pm

- Ensemble Prediction of the Impact of Missense Variants Substantially Decreases VUS Rate in Genetic Testing (R Kueffner, MJ Guillen Sacoto, G Stolovitzky)

Rapid Fire Poster Presentation: Thursday, March 14 from 12:00pm – 12:30pm

- (P130) Phenotypic Spectrum of Individuals with *SLC16A2* Variants: Allan-Herndon-Dudley Syndrome (K McWalter, HZ Elloumi)

Poster Sessions:

Thursday, March 14 from 10:30am – 12:00pm

- (P149) Exome Sequencing vs Chromosomal Microarray for Copy Number Variant Detection (K McWalter, J Juusola, P Kruszka, A Lindy, G Douglas)
- (P293) Long Read Sequencing Analysis of 120 Samples with Known and Challenging-to-Detect Clinical Variants (J Devaney, J Noya, A Berlyoung, A Johnson, J Spangler, R Brandon, KS Hruska, L Lochovsky, P Kruszka, G Stolovitzky, S Newman)
- (P635) Long Read Sequencing for the Analysis of Repeat Expansions Disorders (A Berlyoung, J Noya, A Johnson, J Spangler, R Brandon, KS Hruska, L Lochovsky, J Chin, G Stolovitzky, S Newman, J Devaney)

Friday, March 15 from 10:30am – 12:00pm

- (P130) Phenotypic Spectrum of Individuals with *SLC16A2* Variants: Allan-Herndon-Dudley Syndrome (K McWalter, HZ Elloumi)
- (P180) Expanding the Phenotype Spectrum of Mendelian Diseases with a Genotype-First Approach (MJ Guillen Sacoto,

Houda Elloumi, S McGee, V Ustach, T Brandt, P Kruszka)

- (P640) Amplification and Long-Read Sequencing of the Mitochondrial Genome (J Spangler, A Johnson, J Noya, S Yusuff, R Brandon, KS Hruska, L Lochovsky, J Chin, G Stolovitzky, S Newman, J Devaney)

Industry Symposium Session: Thursday, March 14 from 11:30am – 12:45pm

- Equitable Access to Exomes and Genomes. Ordered by Whom?

ACMG 2024 meeting attendees can visit GeneDx at booth #701

About GeneDx
At GeneDx (Nasdaq: WGS), we believe that everyone deserves personalized, targeted medical care—and that it all begins with a genetic diagnosis. Fueled by one of the world's largest rare disease data sets, our industry-leading exome and genome tests translate complex genomic data into clinical answers that unlock personalized health plans, accelerate drug discovery, and improve health system efficiencies. It all starts with a single test. For more information, please visit **www.genedx.com** and connect with us on **LinkedIn**, **Facebook**, **X**, and **Instagram.**

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1. Srivastava S, Love-Nichols JA, Dies KA, *et al.* Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. *Genet Med.* 2019 Nov;21(11):2413–2421; https://doi.org/10.1038/s41436-019-0554-6
2. Clark MM, Stark Z, Farnaes L, *et al.* Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. *NPJ Genom Med*. 2018 Jul 9;3:16. doi: 10.1038/s41525-018-0053-8.
3. Internal Data